MRCPCH Clinical: Short Cases, History Taking and Communication Skills

Fourth Edition

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Pastest

CONTENTS

Part II

The communication skills station

4

Introduction and Suggested Approach

This section of the exam is intended to look at how you communicate with patients and/or their parents in your everyday clinical practice. You are awarded marks for both your communication skills and clinical knowledge, and so it is not enough (although is essential) to just be a good listener – your job is to address patient's and their family's concerns appropriately and to provide them with reliable, accurate information about the child's condition and the ongoing management plan. This requires a broad knowledge of common paediatric conditions similar to that required in the written part of the MRCPCH exam. In preparing for this station you should concentrate as much on your background knowledge as you do on your communication style.

It is worth reading through the RCPCH Anchor Statements as well as the mark sheets (on the RCPCH website) as you prepare for this section of the exam.

You are being marked in three areas:

1. Conduct of interview
2. Appropriate explanation and negotiation
3. Accuracy of information given.

The six main patterns of communication scenario described by the College are:

1. Information giving
2. Breaking bad news
3. Consent
4. Critical incident

5. Ethics
6. Education.

Here are some suggestions for how to prepare for this section:

1. Clear guidance about expectations and some comprehensive examples of cases are available from the RCPCH (www.rcpch.ac.uk) and reviewing this information should be your first step.
2. Make a shortlist of conditions that you plan to know in detail (see Chapter 6 'Further Scenarios'). You should know about the usual treatments offered and practise how you would talk through these clearly and without using any medical jargon. No conditions should be considered too serious or too trivial to come up in the exam.
3. A good way of practising suggested approaches would be to ask a colleagues to watch you explain management decisions to the parents during a ward round. Give them a copy of the Anchor Statements for the communication skills section. Ask for feedback and suggestions for improvement. Involve parents in the feedback process if you feel comfortable doing this.
4. Role play with colleagues who are also preparing for the exam. Prepare scenarios for each other. Make sure you practise the scenarios you feel least comfortable with. Practise under 'formal' conditions and practise keeping to time.

SUGGESTED APPROACH

All of the scenarios will fit into one of the following broad categories. This is not intended to be a syllabus for studying for the communication skills section, but a framework (after initial introductions) to structure your approach to these stations. This should make you feel much more confident in the run-up to the examination.

Information giving

Talking about chronic conditions

Frequently this takes the form of talking to a child or parent about a change in management because of a clinical deterioration. Parents and or children may disagree with the medical team's interpretation or plan. When parents or patients disagree with the team or need more convincing, a suggested approach is to:

- Ask them what their concerns are

- Discuss each concern: refer to any evidence you know that backs up a sensible medical plan (which could be published articles or national guidelines) or describe what is generally accepted practice
- Talk through the risk/benefit of any alternative management strategies
- When there is potential for harm to the child if there is no agreement, then describe plainly what harm could arise, how likely this is to happen and the expected time frame
- If they remain unconvinced, offer to take their concerns back to the team looking after them and arrange another time to discuss the issue further
- Summarise what you have talked about, including concerns that have been raised and what was decided.

Explaining a new treatment

- Check patients' and parents' understanding of current status and need for change in management
- Describe what the new treatment is
- Explain how it works (use diagrams wherever possible)
- Explain how it is administered
- Explain what the desired/expected outcome is
- Explain how long the patient will need to take it
- Talk about side-effects, how likely they are, how to recognise them and what to do about them
- Give an opportunity for questions and explore any concerns that the patient or their parents have about the new treatment
- Summarise what you've said and check parents' understanding of this:
    - Discuss how the family can access/give written information where possible.

Breaking bad news

- After initial introductions swiftly move on to talking about new information, ie give the diagnosis
- Establish their prior knowledge of this diagnosis – do they know anyone with this condition
- Talk about the implications of the new information
- Allow space for patient/parent to respond, express feelings, ask questions
- Answer concerns honestly and sincerely
- Talk about what is going to happen now
- Summarise, giving the opportunity for more questions
- Offer to meet again to go through any questions
- Offer written literature, for example contact details for support group if appropriate.

5.1 Case one: breaking bad news – new diagnosis of Down syndrome

The following is a fully worked-through example of a scenario that is typical of the sorts of cases you will be expected to deal with in the communication skills section. In keeping with the layout that you will encounter in the exam we have included boxed 'instructions to the candidate' as well as those given to the role player and examiner (who has a copy of all three 'instructions'). These are similar to the instructions seen within the specimen scenarios on the RCPCH website, www.rcpch.ac.uk. The intention is that this, and the following scenarios can be used as templates so that you can develop a structured approach to this section of the exam, and become familiar with the level of background knowledge you will need to be fully prepared. This scenario could fit into the category of 'Breaking bad news' but will also test your ability to give information in layman's language and requires a working knowledge of Down syndrome.

Instructions to candidate

- This station is a test of your ability to convey information based on the specific task given.

Timing:
*Total time provided for this station is **9 min**.*

*There are **2 min** before you are called in to read this sheet and prepare yourself. A bell will signal the start of the station and you will be invited into the examination room – take your instruction sheet with you.*

*You will have **9 min** in total for the consultation; a warning will be issued when you have **2 min** left.*

You will not be assessed on the speed at which you communicate; you may find that you run out of time to finish the consultation.

You may take notes.

You will not be asked to examine the patient.

Role

A Specialist Trainee in paediatrics, working in a district general hospital.

Setting

Side room on the postnatal ward.

You are talking to

Mother of Baby Williams, a baby boy born overnight; he is currently almost 10 hours old. You have just examined him in one of the side rooms on the postnatal ward, as your SHO is concerned he may have Down syndrome. You feel that clinically, based on your examination, he does have Down syndrome.

Other information

Baby Williams was born at term following an uneventful pregnancy, his mother is 27 years old and based on the 'triple test' and nuchal thickness USS, she was not at increased risk of having a baby with Down syndrome. This is her and her husband's first baby.

Task

- ❍ To explain the likely diagnosis
- ❍ To explain further investigations required
- ❍ To explain the implications of the diagnosis (ie what is Down syndrome and how it affects children)

You are not expected to gather any further medical history during this consultation.

Role player information

(This is the information that is provided to whoever is playing the role of the parent.)

> *Total time provided for this station is 9 min.*
>
> *There will be a dialogue between you and the candidate only; you will not communicate with the examiner during the 9-min period.*

Role

You are Jenny Williams and have just given birth to your baby Benji who in your eyes is perfect. Up until this point you have had no concerns about Benji. This is your first baby. You have been waiting a long time for this baby – and have had two previous miscarriages.

You have heard of Down syndrome. You don't know anyone who has it, but have seen people on television with it, and don't really have any idea about it apart from the fact that Down's children often have 'special needs' at school.

Your general feelings

You are feeling more and more anxious – you can tell something is very wrong. You want to know what it is and how to make it better. The midwife asked you to call your husband in, but you do not want to wait for him to get in to the hospital and just want the doctor to tell you straight away what is going on.

What to expect from the candidate and what your feelings and *possible* further questions could be

- The candidate should explain to you that they think Benji has Down syndrome.
- The candidate should explain to you how they will confirm the diagnosis (ie by doing a blood test)
- You should express that you do not know anything about Down syndrome
- The candidate should give you a basic explanation of Down syndrome – how it will effect Benji's development, and other associated problems that will need to be screened/tested for.

After the candidate has explained what Down syndrome is your further questions might include

- How will this affect his future (schooling, keeping up with other children)?
- Will he ever get better from this?
- How could this happen when all your screening tests in pregnancy were normal?
- You and your partner are hoping to have other children; will they be affected too? How did he get this condition; did you give it to him?

Examiner information

> *Total time provided for this station is **9 min**.*
>
> *There will be a dialogue between the candidate and the role player only. Please warn the candidate when only **2 min** of the consultation remain. Otherwise do not communicate with candidate or role player during the **9 min** consultation.*

If the candidate finishes early, you should check that they have finished. If so, they should remain in the room until the session has ended.

Guide notes towards Expected Standard

- Explain that based on your examination Benji has many of the features of a baby with Down syndrome
- You will need to do a blood test to confirm the diagnosis

- Appropriate summary of Down syndrome, including its association with congenital heart defects that will need to be screened for.

Suggested plan of attack

Preparation

In this scenario you are expected to give relevant information in an effective way, but you are also breaking bad news. A breaking bad news 'checklist' which could be adapted for this situation is given in the Box. Remember that in real clinical practice when you are about to discuss something with a parent that you suspect will be distressing or sad for them you need to think about the following:

- Environment: private and distraction free
- Interruption free: hand over your bleep, tell your team where you are and what you're doing
- Are the right people there? This includes relevant medical staff (in this case the midwife looking after Benji and his mother) and the parents'/patient's support network. Are you the right person to be talking about this now (should your senior colleagues be involved)? A senior colleague should certainly be aware that you suspect Down syndrome and are having this discussion
- Do you know all the facts: not only about the patient's background and medical history but about the condition you are talking about and how it is managed in your hospital.

Breaking bad news checklist

- Establish common knowledge base
- Talk about new information
- Talk about the implications of the new information
- Allow space for patient/parent to respond: expressing feelings, asking questions
- Answer concerns honestly and sincerely
- Talk about what is going to happen now
- Summarise with opportunity for more questions
- Offer another meeting
- Offer written literature, contact details for support group if appropriate

Note how the 'task' instructions appear brief and relatively straightforward; however there are clearly several components to this task and all of them are being specifically watched for by the examiner. In the preparation time you

should make a quick list of what you feel needs to be said to fully satisfy your task of 'explaining implications'. This could look like this:

1. Establish how much mother already knows (in this case about Down syndrome).
2. Confirmation of diagnosis, describe what the condition is and (very broadly) the clinical features.
3. Discuss associated conditions to be screened for (eg echo for congenital heart disease, hypothyroidism), potential problems/complications.
4. Support groups/useful websites.

Introduction

You might like to start by saying to the examiner that you would have already handed over your bleep and arranged for a midwife to be present with you (before you say anything to the role player).

When you introduce yourself, say where you fit into Benji's care. Establish a common knowledge base.

> *'Good morning Mrs Williams, my name is Dr Simpson. I'm the specialist trainee in the team looking after Benji. Having just examined Benji I feel he has some features which make me think he may have Down syndrome. Have you heard of Down syndrome or know anyone with it?'*

Explanation

Try to give the diagnosis as soon as possible with limited pre-amble. After you have said it, allow some time for it to sink in. Don't be afraid of silence – what seems like a very long awkward pause to you may not seem like a long time to the parent. You will need to find a way of allowing the parent to set the pace of the discussion following this pause so that you can explain the key points on your list of information to be conveyed.

Closing remarks

You need to check that she has understood one or two key points from the consultation and establish a plan for what will happen now.

> *'Mrs Simpson, I realise that I've given you a lot of information and that it is a lot to take in all at once. The main thing to remember is that I think Benji has Down syndrome. This is something that will affect his development as he grows up, though it is difficult to predict to what extent, it is also linked or associated with some other problems that we need to check Benji for.'*

> *'What is going to happen now is that I am going to do Benji's blood tests to confirm the diagnosis. I should have this result within 2 working days. In the meantime I will arrange for Benji to have an echocardiogram which is an ultrasound of his heart. Before going home we should have the results of these and*

also make sure Benji is feeding well. I'll also give you some patient information about Down syndrome including contact details for a website where you'll be able to find some useful information, Members of the team will be available to answer any questions you will have. We will let your GP and health visitor know.'

A useful website to look at both for yourself and to pass on to parents is www.downs-syndrome.org.uk.

7

INTRODUCTION AND SUGGESTED APPROACH

The clinical stations are seen by many candidates as the most challenging part of the examination. However, thorough preparation and honing of clinical examination technique will give you more confidence – even if you encounter a case you may not have seen before.

7.1 Preparation and technique

Preparation and technique are intertwined. Many candidates fail the exam because of poor 'short case' technique, and this is usually down to poor preparation of potential exam scenarios and a lack of awareness of what is expected of them in each station. The well-prepared candidate will give themselves many opportunities to pick up marks, just as the poorly prepared candidate will inevitably drop marks.

In particular, if your examination technique is poor you will fail.

The RCPCH Anchor Statements for 'Expected Standard/Clear Pass' demonstrate that three key areas are assessed:

- **Conduct of examination –** Introduces oneself, puts parent and child at ease. Displays an appropriate level of confidence. Appropriate pace without rushing. Acknowledges child fully and explains intended examination if deemed appropriate. Adjusts language and behaviour to suit age of child.
- **Clinical examination –** Systematic and uncluttered technique. Able to identify clinical signs and interpret their meaning.
- **Discussion with examiners –** Sensible differential diagnosis. Able to suggest a sensible management plan, including investigations. Demonstrates an understanding of impact of findings on patient and family unit.

For example, if you are asked to examine the respiratory system, to pass the exam you will need to do more than just pick up and present findings. It may be you will need to present not only the most likely diagnosis, eg cystic fibrosis, but also suggest the different investigations that could have led to this diagnosis, such as the sweat test, genetic analysis or the Guthrie test looking at immunoreactive trypsin. This reiterates the philosophy that the exam reflects everyday clinical practice.

- Examine the child
- Pick up and present the abnormalities
- Answer questions relevant to the case

It is essential to be completely clear about how to examine any system. Decide on your own method, learn it and practise it. Use this method throughout the preparation period when seeing short cases. Try not to change your approach in the run-up to the exam. Remember there is no definitive way to examine any system; you need to find your own scheme. Whichever scheme you use, remember that the examiner is unlikely to tolerate imprecise or clumsy clinical examination, and this should be avoidable with proper preparation. This book suggests an examination scheme for most potential scenarios. You are also referred to standard texts of paediatric examination, and up-to-date information regarding the examination from the Royal College of Paediatrics and Child Health website (www.rcpch.ac.uk).

Remember that confidence in the exam comes from knowing that your examination technique is not only correct but is also **reproducible in a highly stressful environment**. This comes from relentless practice of examination and presentation in the company of several different 'examiners'. It is important to be mock-examined by a range of colleagues, some of whom you know and some of whom you don't. This reflects the six different examiners who will soon be assessing you in the clinical stations with no prior knowledge of your

performance in a previous station, or indeed your performance in the workplace. At the very least you should go around in pairs and be very objective when assessing each other's performances. It will help to take a copy of the Anchor Statements with you to see how you measure up to the Expected Standard.

During the examination itself there are several common pitfalls:

- Poor engagement with the child, including failure to look at a child to judge whether you may be eliciting pain. For example, repeated examination of an enlarged spleen may start to become uncomfortable for a patient if you are the fourth consecutive candidate to have seen them. It is important to be empathetic with the patient, just as you would be on the wards.
- Failure to talk your way through an examination when it has been requested/is appropriate.
- Lack of conviction in your findings, enabling an examiner to place doubt in your mind and throw you off balance. For example, a candidate was asked to examine an abdomen with bilateral masses and then asked to present her findings. She presented the patient as having an enlarged spleen and liver. In the subsequent discussion the examiner was able to change the candidate's mind twice as to whether the masses were bilaterally enlarged kidneys or a large liver and spleen.
- Stopping the examination when you have identified the organ in question. For example, there is a set routine for examining an enlarged liver and you must continue with your examination until you are stopped. Another candidate was asked to examine the abdomen. There was an enlarged liver and the child had a fever, having just returned from Africa. The candidate correctly identified malaria as the diagnosis but failed the short case. On counselling she was told that her examination of the liver was inadequate because she did not percuss its size.
- Excluding a clear physical sign if you feel it does not fit in with other signs. Another candidate was asked to examine the cardiovascular system and to comment afterwards. She correctly identified the missing left brachial pulse and the left thoracotomy scar, making the correct diagnosis of repaired aortic coarctation. However, this candidate also heard a soft ejection systolic murmur and could not fit it into the picture and did not mention it. Of course, such a murmur is to be expected in coarctation. Despite this the candidate passed.
- Failing to come up with some common differential diagnoses in the discussion. The differential diagnoses in this text are 'practical differential diagnoses' focusing primarily on common scenarios and ones that are likely to be seen in the exam. Many candidates report that they are not given the chance to present a full differential diagnosis, but are asked for one or two possibilities relating to the child they have seen. Where relevant, the most likely differentials will be at the top of the list in this book.

Basic format for clinical examination

- Introduce yourself to the child and parent
- Be prepared to talk about your findings as you proceed
- Observe and be prepared to comment on:
  - General appearance and health – well or unwell? Ward patients are occasionally used
  - Dysmorphic features – syndromic or otherwise
  - Growth and nutrition – note obvious abnormalities. You may be asked to assess pubertal status
  - Development – neurodevelopmental abnormalities may be noted during examination of other systems. Many patients are eligible for two or more stations per exam session, for example neurology and child development
  - Hands – colour, clubbing, nail abnormalities, poor perfusion
  - Face – cyanosis, anaemia, jaundice
  - General observations such as presence of a nasogastric tube
- Palpate
- Auscultate
- Ask for permission to examine any other relevant part of the body
- Describe your findings
- Answer questions about your findings until the bell rings for the end of the station

7.2 Presentation skills

Presentation skills are important. In particular, the first few sentences you say about a certain scenario need to be practised. Examples of how you should present cases are included in Chapters 8–13. When presenting, it is useful to have a few general words and phrases that can apply to most cases, eg ‘Harry is a boy who looks well grown and I would like to plot his weight on a centile chart …’. This approach overcomes nerves and gives you time to compose yourself.

The information in this section of the book is presented partly in note form and partly as it would appear in the exam, using case scenarios to illustrate the knowledge required on a particular topic. Additional information pertaining to many of the conditions seen in this section can be found in Parts I and II.

11

NEUROLOGICAL/ NEURODISABILITY

CONTENTS

11.3 Cases and conditions: cranial nerves and eyes

Ptosis

This is a very common short case and you will be invited either to comment on or examine the child's eyes.

Instructions to candidate

Examine this 12-year-old boy with ptosis.

Case

This young man has a left ptosis. He is a well-looking boy and there are no dysmorphic features. I would like to examine his eyes. His eyes look normal. The pupils are symmetrical. There is no obvious squint. I would like to test his vision – candidate gets him to read – vision appears normal in both eyes (tested separately). His visual fields and eye movements are normal. His direct and consensual reflexes and his ability to accommodate are normal.

I suspect therefore that his ptosis is congenital.

Notes on ptosis

The examiner will expect you to examine the eyes carefully and in doing so, come up with a likely cause of the ptosis. The ptosis will become apparent on inspection. At this stage, on noting the ptosis, you need to have a clear differential diagnosis in your mind.

Ptosis may be broadly classified as syndromic or neurological in origin.

Most likely in exam:

- Horner syndrome
- Third nerve palsy
- Myasthenia gravis
- Congenital idiopathic ptosis

Full differential diagnosis of ptosis

Syndromic

- Noonan syndrome
- Rubinstein–Taybi (microcephaly, broad thumbs and toes, significant learning difficulties)
- Smith–Lemli–Opitz syndrome
- Marcus Gunn (aberrant innervation causes the affected lid to raise and wink when the child chews or cries)

Neurological

- Third nerve palsy
- Horner syndrome (see below)
- Myasthenia gravis
- Dystrophia myotonica
- Craniosynostosis
- Neuroblastoma
- Migraine

Others

- Congenital idiopathic cause (evaluate early to prevent amblyopia)
- Ocular tumours (rhabdomyosarcoma)

Notes on Horner syndrome

Lesion of the sympathetic nervous system in association with the brainstem or cervical cord or a lesion of the sympathetic plexus in association with the carotid artery.

Features are:

- Partial ptosis
- Pupil constriction
- Enophthalmos
- Ipsilateral anhidrosis
- Heterochromia iridis (complete when one iris is a different colour from the other; partial when part of one iris is a different colour from the rest) – indicates congenital Horner syndrome
- Normal direct and consensual reflex

Squint

Examination of a squint (see Box) is commonly asked for in the exam and needs to be practised carefully. It is best to get an orthoptist to help you to

perfect your technique. Beware of epicanthic folds or hypertelorism giving a false impression of squint.

Examination of squint

- Check the child can see first – with both eyes!
- Corneal reflections
- Eye movements
- Cover test

Notes on squint examination

If you are told that a child has a squint, move straight on to testing after checking the acuity. If you notice a squint during a full eye examination, offer to test the child first without and then with their glasses. The child needs to be co-operative for this to work.

Corneal reflections

Observe both eyes while shining a point source of light at both corneas (corneal reflections). The light source should be held about 30 cm from the eyes and in different planes. If the eyes are straight, the reflection from both corneas will be symmetrical. If a squint is present, the reflection will be asymmetrical.

Eye movements

Eye movements should be examined in all directions using an interesting object. If the eyes deviate, ie there is asymmetry, then a squint is present.

If the deviation is worse in one direction, the squint is paralytic and is caused by a palsy of one of cranial nerves III, IV or VI (for the features of each lesion, see Box). Trauma and tumours are the commonest causes of acquired paralytic squint, and more than one cranial nerve may be affected.

If deviation is the same in all directions, the squint is non-paralytic.

Features of a third (oculomotor) nerve lesion

- Complete ptosis
- Diplopia
- Downward and lateral gaze (unopposed lateral rectus and superior oblique muscles)
- Pupil dilatation
- Failure of the pupil to react to light or to accommodate

Features of a fourth (trochlear) nerve lesion

- Diplopia
- Upward deviation of the eye, +/– head tilt towards unaffected eye (often subtle)
- Failure of downward and lateral gaze (failure of the superior oblique muscle) – difficult to read

Features of a sixth (abducens) nerve lesion

- Diplopia
- Unable to abduct on affected side (failure of the lateral rectus muscle), leading to a convergent paralytic squint

The following tests should be done at near and far vision, first with glasses and then without. This avoids missing a squint caused by excessive adduction secondary to hypermetropia.

Inward deviation is denoted by **'eso'**.

Outward deviation is denoted by **'exo'**.

Cover/uncover test

This examines for a manifest squint (tropia). You will note an asymmetrical corneal light reflection. Ask the child to fix on an interesting object straight ahead at 1 m and then 6 m. Cover the eye that appears to be fixing. If the other eye takes up fixation then there is a manifest squint present.

Alternate cover test

Again, ask the child to fixate on an interesting object at 1 m and then 6 m. Rapidly alternate covering/uncovering between left and right eyes. Look at each eye as it is just uncovered: if one moves to take up fixation, a latent squint (phoria) is present on that side.

Non-paralytic squint

Most squints will be **non-paralytic**. You may be asked about treatment of non-paralytic squint. There are two main aims. The first is to achieve the best possible vision in each eye. Any underlying defects such as cataracts or refractive errors are corrected, and amblyopia is prevented with occlusion therapy. Secondly, achieve the best possible ocular alignment.

Surgery is commonly required.

Nystagmus

Nystagmus describes involuntary, abnormal eye movements that are rhythmic and have either a slow and fast phase (jerky) or are slow in both directions (pendular). Most causes are pathological, but note that some children have nystagmus at the extremes of lateral gaze, which is physiological. This can be more pronounced when the child is tired.

Instructions to candidate

Look at this boy's eyes.

Case

Jack is a well-looking 10-year-old boy who has pale skin, blue eyes and fair hair. Examination of his eyes reveals that he usually wears sunglasses, there is nystagmus and reduced visual acuity. Red reflexes are easily visible. I suspect he has oculocutaneous albinism.

What is the inheritance?

- Autosomal recessive (ocular albinism alone is X-linked recessive)

Notes on oculocutaneous albinism

- Commonest form of albinism; there are four different subtypes
- In all types there is an absolute or relative lack of melanin pigment in eyes, hair and skin
- Opthalmological problems include photophobia, strabismus, reduced acuity as well as nystagmus
- The principal long-term consequences are blindness and skin cancer. Avoidance of sunlight, where practicable, and use of high-factor sunscreens, are essential
- A differential diagnosis of oculocutaneous albinism is Chediak–Higashi syndrome, a rare condition in which there is increased susceptibility to bacterial infections as well as hypopigmented eyes, skin and hair.

Notes on nystagmus

- **Cerebellar nystagmus** is the most important type to recognise. It is jerky and is usually horizontal, and worsens on looking to the side of the lesion. The fast (return) component is directed towards the side of the lesion
- **Vestibular nystagmus** differs in that the slow phase is directed towards the side of the lesion
- **Vertical nystagmus** is usually due to a lesion of the brainstem at the pontomedullary junction (roughly at the foramen magnum), eg achondroplasia, or Arnold–Chiari malformations. It can also be caused by phenytoin or carbamazepine toxicity.
- **Ocular nystagmus** with slow, searching movements occurs in blindness
- **Congenital nystagmus** is present at birth, although it is often not fully recognised until after the first few months. It is usually bilateral and horizontal. If it is idiopathic it often improves with age, although it is important to be aware that there are some more serious causes which carry a poorer prognosis

Fundoscopic findings – notes on various signs

Papilloedema

The stages of papilloedema are:

1. The optic nerve becomes hyperaemic
2. Small capillaries on the optic disc disappear as they become compressed
3. Venous pulsations are no longer seen as the intracranial pressure increases
4. The border of the optic disc becomes indistinct
5. Flame-shaped haemorrhage appears around optic disc

Florid papilloedema is a late sign of raised intracranial pressure.

Causes

- Raised intracranial pressure
- Brain tumours – craniopharyngioma, brainstem glioma, astrocytoma, medulloblastoma
- Leukaemia
- Idiopathic intracranial hypertension (IIH)
- Hydrocephalus
- Systemic hypertension
- CNS infection
- Encephalopathy
- Intracranial haemorrhage
- Craniosynostosis

Cherry red spot – causes

This occurs in certain lysosomal storage diseases.

Seen at the macula, this is the comparatively reddened appearance when one can see through the fovea (which does not build up storage material as relatively less cells) through to reddened choroid below.

This area will be 'cherry red' as it is contrasted with the surrounding retina, which will be pale with build-up of storage material.

Such conditions are:

- Tay–Sachs' disease
- Niemann–Pick's disease
- GM1 gangliosidosis
- Sandhoff's disease
- Mucolipidosis

Lens dislocation – causes

- **Marfan syndrome** – lens dislocation is present in 80% of adults and 50% of 5 year olds with Marfan syndrome. It usually dislocates superiorly and temporally. Symptoms include blurred vision (due to refractive changes) and diplopia. If the pupil is dilated, the edge of the lens may be seen as a black crescent on ophthalmoscopic examination. A clearer view is obtained with slit lamp examination; mention this in the exam
- Homocystinuria – lens dislocates downwards and nasally
- Ehlers–Danlos syndrome

Cataracts – causes

- Metabolic disease, eg galactosaemia

- Wilson's disease
- Hypoparathyroidism
- Diabetes
- Dystrophica myotonica
- Intrauterine infections
- Traumatic insult to the eye

Corneal clouding – causes

- GM1 gangliosidosis
- Fucosidosis
- Mucopolysaccharidosis
- Mucolipidosis

Systemic conditions with eye signs

Condition	Sign
Neurofibromatosis	Lisch nodule, ptosis
Wilson's disease	Kayser–Fleischer ring
Ataxia telangiectasia	Conjunctival telangiectasia
Down syndrome	Brushfield spots – not pathognomonic
Hyperthyroidism	Exophthalmos, lid lag
Juvenile idiopathic arthritis	Iridocyclitis

Table 11.2: Systemic conditions with eye signs

Miscellaneous eye problems

Coloboma

Sometimes seen in the examination. Either inherited (autosomal dominant) or as part of the conditions listed below:

- Aniridia–Wilms' association
- CHARGE association
- Goldenhar syndrome

Duane syndrome

A congenital abnormality of innervation causing simultaneous contraction of medial and lateral recti on attempted adduction of the affected eye. On examination they may have exotropia or esotropia, and on testing eye movements they may have failure to abduct or adduct the affected eye.

Moebius syndrome

This is manifest as a (usually) bilateral but asymmetrical lower motor neurone facial nerve palsy, associated particularly with sixth nerve palsies. These cause failure of abduction and paralytic convergent squints of the affected eyes. The cause is unknown, but as both cranial nerves are closely related it may be due to underdevelopment of the cranial nerve nuclei.

Uveitis

This is inflammation of the uveal tract consisting of the inner vascular coat of the eye, the iris, the ciliary body and choroid. Iritis may occur alone or in conjunction with the ciliary body as iridocyclitis. This commonly occurs insidiously without symptoms.

Children with seropositive (ANA) arthritis are in a high-risk group for uveitis and need to have regular eye reviews.

Case

Jane is a well-looking 8-year-old girl. She appears cushingoid. Examination of her eyes reveals conjunctival injection. The visual acuity of her left eye is markedly reduced. It was not possible to examine the visual fields. The pupil responses are present but slow on the left. Ophthalmoscopy reveals clouding of the cornea on the left. There is normal visual acuity in her right eye with normal pupil responses and no clouding of the cornea.

This would fit with chronic uveitis. I would like to proceed to a full systemic examination, paying particular attention to the joints and looking for the side-effects of steroids.